

of these Databases are eagerly awaited. Version 3.0 of the London Dysmorphology Database (LDDb), London Neurogenetics Database (LNDB), and Dysmorphology Photo Library has been completely updated. This version of LDDb contains information on 3428 dysmorphic syndromes with over 33 000 references. The new version of LNDB has also been expanded and covers 3292 neurogenetic disorders and syndromes involving the central and peripheral nervous systems with over 36 000 references. Version 3.0 of the Dysmorphology Photo Library contains 12 742 photographs, 1355 more than the previous version.

Both Databases and the Photo Library are included on a single CD-ROM. Minimum system requirements for the package include PC with Pentium processor and CD-ROM drive, 32 MB RAM, 150 MB free hard disk space, VGA monitor with 800 × 600 screen resolution and, Internet Explorer 4.0. The software is compatible with all versions of Windows from Windows 95 onwards (including the recently introduced Windows XP). The Databases and Photo Library were easy to install and worked well with Windows Millennium Edition.

Like the previous versions, this version is an authoritative source of information about dysmorphic and neurogenetic syndromes. Although LDDb mainly contains information about non-chromosomal multiple congenital anomaly syndromes, it also includes information about a few distinctive chromosomal deletion or microdeletion syndromes and syndromes resulting from uniparental disomy of different chromosomes. Both Databases have a detailed record for each syndrome with information about its chromosomal location (if known or relevant), McKusick number, synonyms, gene symbol, inheritance pattern, an abstract card, a features card, references card, and photo card. The abstract card provides a compact but informative review of the syndrome. The features card contains an exhaustive list of clinical features and the references card lists all the key published references for that syndrome. The photo card is a superb collection of photographs that show the main facial dysmorphic features of the syndrome and other relevant images, such as skeletal survey, hair microscopy, etc. In the case of neurogenetic syndromes, the photo card contains CT and MRI images showing their characteristic neuroradiological features, examples of EEG changes or the changes observed on other key electrophysiological investigations and, where relevant, pictures of the characteristic neuropathology (including nerve and muscle biopsy).

One of the most useful functions of these Databases is their ability to allow the user to search for syndromes based on the features of a patient. In LDDb these include mainly clinical and/or radiological features, whereas in LNDB a search can be carried out using age at onset of neurological features, neurological and other clinical features, neuroradiological findings, changes seen on electrophysiological investigations, abnormal biochemistry, and neuropathological findings. Syndromes can also be searched for by McKusick number, inheritance pattern, or by references. References can be searched for by their title, author(s), journal, year of publication, or publisher.

An important feature new to this version of the Databases is a new look user interface with a user friendly toolbar. For users who are familiar with version 2.0 of these Databases, it

is possible to configure the display to the style of the previous version with buttons instead of the toolbar. Other novel features of this version include major changes in the photo card and the photograph viewer. These include the ability to change the size of the thumbnails of the images in the Photo Library, display several or all images for a syndrome simultaneously for comparison, and import selected images of a syndrome from the Photo Library to a personal collection of images called "My Collection". It is also possible for users to import clinical photographs of their own patients into "My Collection", thus allowing comparisons to be made with the images in the Photo Library. Most picture file formats are supported, such as JPEG, Windows Bitmap, and Tagged Image File Format, making this a very useful function. There are also links from the Databases to Online Mendelian Inheritance in Man (OMIM) and Medline in case the user wishes to obtain more information about an individual syndrome or reference.

Another important feature of version 3.0 is the ability to download regular updates to both the LDDb and LNDB online, at no extra cost. This will allow the Databases to remain up to date with all recent advances, especially with respect to identification of genes for well recognised syndromes and delineation of new dysmorphic and neurogenetic syndromes.

Both Databases are relatively easy to use, even for first time users. However, a working knowledge of clinical dysmorphology and neurology is necessary to harness their full potential and in order for these Databases to be used effectively. It is difficult to find fault with any aspect of the Databases. The only drawback is the price of the combined package (£1595 + VAT for the single user version and £1125 + VAT for the single user upgrade). Although the Databases and Photo Library can all be purchased individually, the cost of each component (£595 + VAT for the single user version and £425 + VAT for the single user upgrade) is still prohibitive and will dissuade most clinicians from buying a copy for their personal use.

The clinical genetics community owes a huge debt of gratitude to the two authors of these Databases. It is difficult to envisage the practice of clinical dysmorphology and neurogenetics without access to the LDDb, LNDB, and Dysmorphology Photo Library. For most clinical geneticists faced with an unknown dysmorphic or neurogenetic syndrome, these Databases are likely to be the first port of call. A thorough search of the Databases will often suggest a diagnosis for such patients. It is this reviewer's firm belief that no clinical genetics or neurology department can afford to be without these Databases. Perinatal pathologists and paediatricians will also benefit enormously from access to them.

Mohnish Suri

Fragile X Syndrome - Diagnosis, Treatment and Research

Third edition. Editors Randi Jenssen Hagerman, Paul J Hagerman. £65.50 HB, £31.00 PB. Baltimore: The Johns Hopkins University Press. 2002. ISBN 0-8018-6844-0.

This is the third edition of a book that is already well known to clinical geneticists and genetic counsellors. Probably, just as many

scientists from the cytogenetic and DNA laboratories are familiar with the title. The first edition was printed in 1991, at the same time that the mutation that causes fragile X syndrome was identified. When the second edition appeared in 1996, much more could be written about the gene product, FMRP. Advances in understanding molecular and cellular changes in fragile X syndrome have proceeded, but in smaller increments in more specialised fields. The neuroscientific aspects of the syndrome are thus allotted more space in this new edition and, perhaps related to this, there has been an editor substitution with Paul Hagerman, spouse of Randi and molecular arm of the husband-wife collaboration, replacing Amy Cronister, who continues to make a major contribution co-authoring the chapter on genetic counselling.

To make room for new information on neuroscience, the chapter on cytogenetics of fragile X syndrome has been cut but, really, this omission should be regarded as a sound reason for retaining the second edition on the bookshelf. Happily, the opening chapter by Randi Hagerman is not shortened as it contains a wealth of clinical information including perfect illustrations of macroorchidism and the value of a Prader orchidometer. Older geneticists may wistfully recall epic estimations of testicular volume in burly men pre-1991, but new trainees need only gaze in astonishment at these pictures and perhaps give silent thanks for the trinucleotide repeat.

The essays in this edition also provide a good illustration of how one dramatic discovery, the CGG expansion, clarifies much that was previously perplexing, but also leads to many more complex questions being posed. Ted Brown's very clear chapter on the molecular biology of the fragile X mutation sits alongside Stephanie Sherman's fascinating account of the syndrome's epidemiology, but self-congratulatory feelings about one's adult learning capacity are then dashed by detailed state of play reviews of protein studies, an animal model, and the brain structural phenotype. After this, a review of the neuropsychology of the syndrome brought home to me the importance of a technical approach to the definition of behavioural phenotypes, and wound up the first half of the book.

In the second half, essays explore treatment options and give practical advice on dealing with learning difficulties and troublesome behaviours. Much of the best advice is of a general nature and is not specific to the management of children with fragile X syndrome. The chapter on drug therapies will also be of interest to the paediatrician or specialist in learning disability who supervises clinical management. The accounts of academic and psychological interventions that may improve quality of life for affected subjects and their families contain much information that is of value to teachers and therapists. To close, there are useful appendices with web and e-mail addresses for general information, educational software, and other helpful resources.

In summary, this is a book that may be read by the fireside; especially pleasing are the many paragraphs that supply historical background to major discoveries about the syndrome, but the book's main use is for reference purposes. Quite simply, it should always be consulted when considering clinical problems.

John Tolmie